

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

**Table 3. CAV3 Mutations and Associated Phenotypes**

Nt Change	Exon	Protein	Position	Phenotype	Reference
77G>A	1	R26Q	N-terminal	Isolated HCK (s)	Carbone, 2000
"	"	"	"	RMD	Vorgerd, 2001 Schara, 2002
"	"	"	"	LGMD-1	Figarella-Branger 2003
"	"	"	"	DM	Tateyama 2002
"	"	"	"	RMD + DM	Yabe 2002
"	"	"	"	LGMD-1C + RMD	Fee 2004
"	"	"	"	Isolated H-CK	Fulizio 2005
81C>A	1	D27E	N-terminal	LGMD-1C + RMD + DM	Fisher 2003
82C>A	1	P28T	N-terminal	RMD	Van der Bergh, 2004
83C>T	1	P28L	N-terminal	Isolated HCK (f)	Merlini, 2002
96C>G	1	N32K	N-terminal	LGMD-1C	Sugie 2004
99C>G	1	N33K	N-terminal	DM+ HCK	Fulizio 2005
IVS+2T>C	1	No protein	N-terminal	LGMD-1C	Muller 2006
128T>A	2	V43E	N-terminal	LGMD-1C	Sugie 2004
133G>A	2	A45T	N-terminal	LGMD-1C	Herrmann 2000
"	"	"	"	RMD	Betz 2001
"	"	"	"	LGMD-1C+ HCK	Fulizio 2005
134C>T	2	A45V	N-terminal	RMD	Betz 2001
136G>A	2	E46K	N-terminal	RMD	Ricardo 2005
154A>G	2	S52G	Scaffolding domain	RMD	Dotti2006
169G>A	2	V57M	Scaffolding domain	Isolated HCK (f)	Alias 2004
183C>A	2	S61R	Scaffolding domain	LGMD-1C+ HCK	Fulizio 2005
187A>C	2	T63P	Scaffolding domain	LGMD-1C	Matsuda 2001
188C>G	2	T63S	Scaffolding domain	HCM	Hayashi 2004
186-194del	2	TFT63- 65del	Scaffolding domain	LGMD-1C	Minetti 1998
233C>T	2	T78M	Membrane- spanning	LQTS	Reijneveld 2006 Vatta 2006

Nt Change	Exon	Protein	Position	Phenotype	Reference
			domain		
257T>C	2	L85P*	Membrane-spanning domain	RMD	Kubish 2003
277G>A	2	A92T*	Membrane-spanning domain	RMD + LGMD-1C	Kubish 2003
290T>G		F96C	Membrane-spanning domain	LQTS	Vatta 2006
290-293del	2	F96del	Membrane-spanning domain	LGMD-1C + RMD + HCK	Cagliani 2003
314C>T	2	P104L	Membrane-spanning domain	LGMD-1C	Minetti 1998
423C>G	2	S140R	C-terminal domain	LQTS	Vatta 2006

Taken from <http://www.hgmd.cf.ac.uk/ac/index.php>

HCK, hyperCKemia, (s) sporadic, (f) familiar

RMD, rippling muscle disease

LGMD, limb-girdle muscular dystrophy

DM, distal myopathy

HCM, hypertrophic cardiomyopathy

LQTS, long QT syndrome

\*, homozygous mutation

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